

# Association between Fat Mass and Obesity Associated (FTO) Gene Polymorphism (rs9939609) and Polycystic Ovary Syndrome (PCOS): A Sri Lankan Study

Branavan Umayal<sup>1</sup>, Muneeswaran Kajan<sup>2</sup>, N.V. Chandrasekharan<sup>2</sup>, W.S.S. Wijesundera<sup>3</sup> and Chandrika N. Wijeyaratne\*<sup>1</sup>

<sup>1</sup>Department of Obstetrics and Gynecology, Faculty of Medicine, University of Colombo, Sri Lanka

<sup>2</sup>Department of Chemistry, Faculty of Science, University of Colombo, Sri Lanka;

<sup>3</sup>Department of Biochemistry and Molecular Biology, Faculty of Medicine, University of Colombo, Sri Lanka.

(e-mail - [mandika59@hotmail.com](mailto:mandika59@hotmail.com))

**Abstract: Objective:** to study the association of FTO gene rs9939609 variant in Sri Lankan women with PCOS.

**Method:** Consecutive women with PCOS (n=55) and controls (n=110) were recruited. Clinical parameters, anthropometry and biochemistry were measured in all. DNA was extracted and tetra ARMS PCR was carried out for genotyping. The association between rs9939609 variant of FTO gene and PCOS susceptibility was examined.

**Results:** High frequency of A allele was observed in PCOS group. The frequency of T allele (normal allele) was significantly higher in controls (PCOS - AA= 40%, AT=23.6%, TT=36.3%; Controls AA = 13.6%, AT=20.9%, TT=65.4%). Significant correlation has been found between FTO gene and BMI (chi square value = 17.05, p<0.05).

**Conclusion:** The rs9939609 variant of FTO gene is associated with PCOS susceptibility in Sri Lankan women, probably because of its effect on body mass index (BMI).

**Keywords:** Fatty mass and obesity associated (FTO) gene, Poly Cystic Ovary Syndrome (PCOS), Body Mass Index (BMI)

## 1. Introduction

Polycystic ovary syndrome (PCOS) is the commonest endocrine disorder in women of reproductive age (Balen *et al.*, 1995). Diagnosis of PCOS is based on the 'Rotterdam criteria', which require the presence of two of the three following features: polycystic ovaries, anovulation and androgen excess (clinical and or biochemical). Genome-wide association study identified the fat mass and obesity-associated (FTO) gene as an obesity susceptibility gene (Frayling *et al.*, 2007). The human FTO gene is located on chromosome 16 and expressed in a wide range of tissues, including the adipose tissue and specific areas of the brain and muscles, suggesting its potential role in body weight regulation (Wehr *et al.*, 2010). Several single nucleotide polymorphisms (SNP) of the FTO gene have been described. The variant FTO rs9939609 is the most extensively studied, located within the first FTO intron which has two alleles, A and T, the former has been linked to an increased risk for both obesity and type 2 diabetes mellitus (De Luis *et al.*, 2013).

Given the high prevalence of obesity in women with PCOS, we hypothesized obesity susceptibility gene may play an important role in the development of PCOS. In this study, we aimed to identify the association of FTO gene variant rs9939609 with PCOS susceptibility in Sri Lankan women with PCOS.









